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UniProtKB/Swiss-Prot entry

O75844

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Entry information

Entry name	FACE1_HUMAN
Primary accession number	O75844
Secondary accession numbers	Q8NDZ8 Q9UBQ2
Integrated into Swiss-Prot on	July 15, 1999
Sequence was last modified on	April 27, 2001 (Sequence version 2)
Annotations were last modified on	September 2, 2008 (Entry version 78)
Name and origin of the protein	
Protein name	CAAX prenyl protease 1 homolog
Synonyms	EC 3.4.24.84 Prenyl protein-specific endoprotease 1 Farnesylated proteins-converting enzyme 1 FACE-1 Zinc metalloproteinase Ste24 homolog
Gene name	Name: ZMPSTE24 Synonyms: FACE1, STE24
From	Homo sapiens (Human) [TaxID: 9606]
Taxonomy	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo.
Protein existence	1: Evidence at protein level;

References

[1] NUCLEOTIDE SEQUENCE [MRNA].

TISSUE=Brain;
DOI=10.1016/S0304-4165(98)00170-6; PubMed=10076063 [NCBI, ExPASy, EBI, Israel, Japan]
Kumagai H., Kawamura Y., Yanagisawa K., Komano H.;
"Identification of a human cDNA encoding a novel protein structurally related to the yeast membrane-associated metalloprotease, Ste24p."
Biochim. Biophys. Acta 1426:468-474(1999).

[2] NUCLEOTIDE SEQUENCE [MRNA].

TISSUE=B-cell, and Fetal brain;

DOI=10.1083/jcb.142.3.635; PubMed=9700155 [NCBI, ExPASy, EBI, Israel, Japan]

Tam A., Nouvet F.J., Fujimura-Kamada K., Slunt H., Sisodia S.S., Michaelis S.;

"Dual roles for Ste24p in yeast a-factor maturation: NH2-terminal proteolysis and COOH-terminal CAAX processing.";

J. Cell Biol. 142:635-649(1998).

[3] NUCLEOTIDE SEQUENCE [MRNA].

TISSUE=Ovary;

DOI=10.1006/geno.1999.5834; PubMed=10373325 [NCBI, ExPASy, EBI, Israel, Japan]

Freije J.M.P., Blay P., Pendas A.M., Cadinanos J., Crespo P., Lopez-Otin C.;

"Identification and chromosomal location of two human genes encoding enzymes potentially involved in proteolytic maturation of farnesylated proteins.";

Genomics 58:270-280(1999).

[4] NUCLEOTIDE SEQUENCE [LARGE SCALE GENOMIC DNA].

DOI=10.1038/nature04727; PubMed=16710414 [NCBI, ExPASy, EBI, Israel, Japan]

Gregory S.G., Barlow K.F., McLay K.E., Kaul R., Swarbreck D., Dunham A., Scott C.E., Howe K.L.,

Woodfine K., Spencer C.C.A., Jones M.C., Gillson C., Searle S., Zhou Y., Kokocinski F., McDonald L.,

Evans R., Phillips K., Atkinson A., Bentley D.R.;

"The DNA sequence and biological annotation of human chromosome 1.";

Nature 441:315-321(2006).

[5] NUCLEOTIDE SEQUENCE [LARGE SCALE MRNA], AND VARIANT ALA-137.

TISSUE=Testis;

DOI=10.1101/gr.2596504; PubMed=15489334 [NCBI, ExPASy, EBI, Israel, Japan]

The MGC Project Team;

"The status, quality, and expansion of the NIH full-length cDNA project: the Mammalian Gene Collection (MGC).";

Genome Res. 14:2121-2127(2004).

[6] VARIANT MADB ARG-340.

DOI=10.1093/hmg/ddg213; PubMed=12913070 [NCBI, ExPASy, EBI, Israel, Japan]

Agarwal A.K., Fryns J.-P., Auchus R.J., Garg A.;

"Zinc metalloproteinase, ZMPSTE24, is mutated in mandibuloacral dysplasia.";

Hum. Mol. Genet. 12:1995-2001(2003).

[7] INVOLVEMENT IN LETHAL TIGHT SKIN CONTRACTURE SYNDROME.

DOI=10.1093/hmg/ddh265; PubMed=15317753 [NCBI, ExPASy, EBI, Israel, Japan]

Navarro C.L., De Sandre-Giovannoli A., Bernard R., Boccaccio I., Boyer A., Genevieve D., Hadj-Rabia

S., Gaudy-Marqueste C., Smitt H.S., Vabres P., Faivre L., Verloes A., Van Essen T., Flori E., Hennekar

R., Beemer F.A., Laurent N., Le Merrer M., Cau P., Levy N.;

"Lamin A and ZMPSTE24 (FACE-1) defects cause nuclear disorganization and identify restrictive dermopathy as a lethal neonatal laminopathy.";

Hum. Mol. Genet. 13:2493-2503(2004).

Comments

- **FUNCTION:** Proteolytically removes the C-terminal three residues of farnesylated proteins. Acts on lamin A/C.
- **CATALYTIC ACTIVITY:** The peptide bond hydrolyzed can be designated -C|-A-A-X in which C is an S-isoprenylated cysteine residue, A is usually aliphatic and X is the C-terminal residue of the substrate protein, and may be any of several amino acids.
- **COFACTOR:** Binds 1 zinc ion per subunit (*By similarity*).
- **SUBCELLULAR LOCATION:** Endoplasmic reticulum membrane; Multi-pass membrane protein. Golgi apparatus membrane; Multi-pass membrane protein (*Probable*).
- **TISSUE SPECIFICITY:** Widely expressed. High levels in kidney, prostate, testis and ovary.
- **DISEASE:** Defects in ZMPSTE24 are the cause of mandibuloacral dysplasia with type B lipodystrophy

(MADB) [MIM:608612]. Mandibuloacral dysplasia (MAD) is a rare autosomal recessive disorder characterized by mandibular and clavicular hypoplasia, acroosteolysis, delayed closure of the cranial suture, joint contractures, and types A or B patterns of lipodystrophy. Type B lipodystrophy observed MADB, is characterized by generalized fat loss.

- **DISEASE:** Defects in ZMPSTE24 are a cause of lethal tight skin contracture syndrome [MIM:275210]; also called restrictive dermopathy (RD). Lethal tight skin contracture syndrome is a rare disorder mainly characterized by intrauterine growth retardation, tight and rigid skin with erosions, prominent superficial vasculature and epidermal hyperkeratosis, facial features (small mouth, small pinched nose and micrognathia), sparse/absent eyelashes and eyebrows, mineralization defects of the skull, thin dysplastic clavicles, pulmonary hypoplasia, multiple joint contractures and an early neonatal lethal course. Liveborn children usually die within the first week of life. The overall prevalence of consanguineous cases suggested an autosomal recessive inheritance.
- **SIMILARITY:** Belongs to the peptidase M48A family [view classification].
- **WEB RESOURCE:** Name=GeneReviews; URL="<http://www.genetests.org/query?gene=ZMPSTE24>"

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Cross-references

Sequence databases

	AB016068; BAA33727.1; -; mRNA.	[EMBL / GenBank / DDBJ] [CoDingSequence]
	AF064867; AAC68866.1; -; mRNA.	[EMBL / GenBank / DDBJ] [CoDingSequence]
EMBL	Y13834; CAB46277.1; -; mRNA.	[EMBL / GenBank / DDBJ] [CoDingSequence]
	AL050341; CAB81610.1; -; Genomic_DNA.	[EMBL / GenBank / DDBJ] [CoDingSequence]
	BC037283; AAH37283.1; -; mRNA.	[EMBL / GenBank / DDBJ] [CoDingSequence]

RefSeq NP_005848.2; -.

UniGene Hs.132642

3D structure databases

ModBase O75844.

Protein-protein interaction databases

IntAct O75844; -.

Protein family/group databases

MEROPS M48.003; -.

Organism-specific databases

H-InvDB HIX0000469; -.

HGNC HGNC:12877; ZMPSTE24.

GenAtlas ZMPSTE24.

HPA HPA006988; -.

MIM 275210; phenotype. [NCBI / EBI]

606480; gene. [NCBI / EBI]

608612; phenotype. [NCBI / EBI]

Orphanet 1662; Dermopathy restrictive lethal.

2457; Mandibuloacral dysplasia.

PharmGKB PA37466; -.

GeneCards O75844.

Gene expression databases

ArrayExpress O75844; -.

CleanEx HS_ZMPSTE24; -.

GermOnline ENSG00000084073; Homo sapiens.

Ontologies

GO:0008235; Molecular function: metalloexopeptidase activity (*traceable author statement from ProtInc*).

GO GO:0006508; Biological process: proteolysis (*traceable author statement from ProtInc*).
QuickGo
view.

Family and domain databases

InterPro IPR006025; Pept_M_Zn_BS.
IPR001915; Peptidase_M48.
Graphical view of domain structure.

Pfam PF01435; Peptidase_M48; 1.
Pfam graphical view of domain structure.

PROSITE PS00142; ZINC_PROTEASE; FALSE_NEG.

BLOCKS O75844.

Proteomic databases

PeptideAtlas O75844; -.

Genome annotation databases

Ensembl ENSG00000084073; Homo sapiens. [Contig view]

GeneID 10269; -.

KEGG hsa:10269; -.

Phylogenomic databases

HOGENOM O75844; -.

HOVERGEN O75844; -.

Other

SOURCE ZMPSTE24; Homo sapiens.

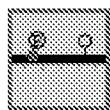
ProtoNet O75844.

UniRef View cluster of proteins with at least 50% / 90% / 100% identity.

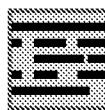
Keywords

Disease mutation; Endoplasmic reticulum; Golgi apparatus; Hydrolase; Membrane; Metal-binding; Metalloprotease; Polymorphism; Protease; Transmembrane; Zinc.

Features



Feature table viewer



Feature aligner

Key	From	To	Length	Description	FTId
CHAIN	1	475	475	CAAX prenyl protease 1 homolog.	PRO_0000138844
TRANSMEM	19	39	21	Potential.	
TRANSMEM	82	102	21	Potential.	
TRANSMEM	124	144	21	Potential.	
TRANSMEM	171	191	21	Potential.	
TRANSMEM	196	216	21	Potential.	
TRANSMEM	348	368	21	Potential.	
TRANSMEM	383	405	23	Potential.	
ACT_SITE	336	336		By similarity.	
ACT_SITE	419	419		Proton donor (By similarity).	
METAL	335	335		Zinc; catalytic (By similarity).	
METAL	339	339		Zinc; catalytic (By similarity).	
METAL	415	415		Zinc; catalytic (By similarity).	
VARIANT	137	137	1	T -> A (in dbSNP:rs17853725 [NCBI]).	VAR_034711
VARIANT	340	340	1	W -> R (in MADB).	VAR_019308

CONFLICT 16 16 E -> K (in Ref. 1; BAA33727).

Sequence information

Length: 475 AA [This is the length of the unprocessed precursor]

Molecular weight: 54813 Da [This is the MW of the unprocessed precursor]

CRC64: 6C49179DEB0C8F7F [This is a checksum on the sequence]

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      10      20      30      40      50      60
MGMWASLDAL WEMPAEKRI F GAVLLFSWTV YLWETFLAQR QRRIYKTTTH VPPELGQIMD

      70      80      90     100     110     120
SETFEKSRLY QLDKSTFSFW SGLYSETEGT LILLFGGIPY LWRLSGRFCG YAGFGPEYEI

     130     140     150     160     170     180
TQSLVFLLLA TLFSALTGLP WSLYNTFVIE EKHGFNQOTL GFFMKDAIKK FVVTQCILLP

     190     200     210     220     230     240
VSSLLLYIIK IGGDYFFIYA WLFTLVVSLV LVTIYADYIA PLFDKFTPLP EGKLKEEIEV

     250     260     270     280     290     300
MAKSIDFPLT KVVVVEGSKR SSHSNAYFYG FFKNKRIVLF DTLLEEYSVL NKDIQEDSGM

     310     320     330     340     350     360
EPRNEEEGNS EEIKAKVKNK KQGCKNEEVL AVLGHELGHW KLGHTVKNII ISQMNSFLCF

     370     380     390     400     410     420
FLFAVLIGRK ELFAAFGFYD SQPTLIGLLI IFQFIFSPYN EVLSFCLTVL SRRFEFQADA


     430     440     450     460     470
FAKKLGKAKD LYSALIKLNK DNLGFPVSDW LFSMWHYSHP PLLERLQALK TMKQH
  
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O75844 in
FASTA format

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 BLAST submission on
ExPASy/SIB
or at NCBI (USA)



Sequence analysis tools: ProtParam, ProtScale,
Compute pI/Mw, PeptideMass, PeptideCutter,
Dotlet (Java)



ScanProsite, MotifScan



Submit a homology modeling request to SWISS-
MODEL



NPSA Sequence analysis
tools




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